What is claimed is:

1. An isolated and purified human MSH5 protein having the amino acid sequence set forth in SEQ ID NO:2, or a fragment of at least six amino acids thereof.

2. An isolated and purified nucleotide segment having the sequence as set forth in SEQ ID NO.:1.

3. An isolated nucleotide segment containing a fragment of at least 17 contiguous nucleotides as set forth in SEQ ID NO:1.

4. An isolated nucleic acid segment having a nucleotide sequence selected from the group consisting of SEQ ID NOs.:3-53.

5. An isolated DNA segment which hybridizes under stringent conditions to a DNA fragment having the nucleotide sequence set forth in SEQ ID NO:1 or a unique fragment thereof and codes for a MSH5 gene.

6. A vector containing the DNA of claim 5.

7. The vector of claim 6, wherein said vector is a retroviral vector.

8. A host transformed with the vector of claim 6 or 7.

9. A vector containing an antisense DNA segment of the nucleotide sequence set forth in SEQ ID NO:1 or a unique fragment thereof.

10. A kit for determining an alteration in a mammalian MSH5 gene by DNA amplification comprising:

a set of DNA oligonucleotide primers in a vial, said set allowing synthesis of a DNA encoding the DNA mismatch repair gene.

11. The kit of claim 10, wherein the DNA mismatch repair gene is hMSH5.

12. The kit of claim 10, wherein said primers are selected from the group of SEQ ID NOs:3-50.

13. A method of determining whether there is an alteration in a mammalian MSH5 gene which comprises:

a) isolating a biological specimen from a preselected mammal;

b) testing the specimen for an alteration in said mammalian MSH5 nucleotide sequence or its expression product; and

c) comparing the results obtained in step b) with a wild type control.

14. The method of claim 13, wherein the biological specimen is selected from blood, tissue, serum, stool, urine, sputum, cerebrospinal fluid, supernatant from cell lysate and a eukaryotic cell sample.

15. The method of claim 13, wherein the mammal is a human.

16. The method of claim 13, wherein an alteration is indicative of a predisposition to malignant growth of cells in the mammal.

17. The method of claim 13, wherein an alteration is indicative of

a predisposition to a malady associated with inappropriate meiotic segregation.

18. The method of claim 15, wherein the biological specimen is selected from a group of blood related individuals.

19. The method of claim 13, wherein the nucleotide sequence is a gene.

20. The method of claim 17, wherein the malady is infertility or Downs Syndrome.

21. The method of claim 13, wherein the expression product is mRNA.

22. The method of claim 13, wherein the expression product is a protein.

23. The method of claim 13, wherein the alteration is in the nucleotide sequence of the DNA.

24. The method of claim 23, wherein the alteration is detected using a method of DNA amplification.

25. The method of claim 24, wherein the method of DNA amplification detects an alteration in at least one intron or exon.

26. The method of claim 25, wherein the alteration is detected in a MSH5 gene using a pair of oligonucleotide primers.

27. The method of claim 25, wherein the wild-type hMSH5 gene has SEQ ID NO:1.

28. The method of claim 13, wherein the alteration is detected by measuring the level of gene expression.

29. The method of claim 13, wherein the alteration is detected by identifying a mismatch between (1) a MSH5 or its mRNA in said tissue and (2) a nucleic acid probe complementary to a mammalian wild-type MSH5, when (1) and (2) hybridize to each other to form a duplex.

30. The method of claim 29, wherein the nucleic acid probe is a DNA probe.

31. The method of claim 29, wherein the mismatch is identified by enzymatic cleavage.

32. The method of claim 13, wherein the alteration in the MSH5 DNA is detected by amplification of MSH5 genes and hybridization of the amplified sequences to nucleic acid probes that are complementary to mutant MSH5 alleles.

33. A method of diagnosing a DNA mismatch repair defective tumor of a mammal, comprising:

isolating a tissue from said mammal suspected of being a tumor; and

detecting an alteration in a MSH5 gene or its expression product, wherein said alteration is indicative of a DNA mismatch repair defective tumor.

34. The method of claim 33, wherein the mammal is a human.

35. The method of claim 34, wherein the DNA mismatch repair defective tumor is lung, breast, colorectal ovary, endometrial (uterine),

renal, bladder, skin, rectal and small bowel.

36. A method of prognosis in an individual having cancer, comprising, comparing a cancer cell from said individual with a non-cancer cell from said individual for the presence of an alteration in the MSH5 gene.

37. The method of claim 36, wherein an alteration in both cells indicates a genetic basis for said cancer.

38. A method of screening for agents affecting a mammalian MSH5 gene comprising:

a) selecting a first test cell having an alteration in the mammalian MSH5 gene;

b) selecting a second test cell, said second cell derived from said first cell, but not having the alteration in the MSH5 DNA;

c) contacting said test cells with a selected agent; and

d) comparing the effects of said agent on the first and second test cells.